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LABORATORY DIAGNOSIS OF SICKLE CELL ANEMIA

Isakulova Muhabbat Mardanovna
Assistant of the Department of Clinical and Laboratory Diagnostics with a Course of Clinical and Laboratory Diagnostics at the Faculty of Postgraduate Education,

Xidoyatullayeva Aziza
Cadet of the Department of Clinical and Laboratory Diagnostics with a Course of Clinical and Laboratory Diagnostics of FOPE,
Samarkand State Medical University, Uzbekistan, Samarkand

Abstract
Sickle cell anemia is one of the most pressing diseases in the field of hemato-oncology, affecting millions of people worldwide. This hereditary disease, caused by a mutation in the gene responsible for the synthesis of beta-globin, leads to a change in the shape of red blood cells, which in turn causes their abnormal aggregation, impeding blood circulation and reducing the level of oxygen in the tissues. The relevance of the problem lies not only in the high prevalence of the disease, but also in the complexity of its diagnosis and treatment. This hereditary disease, caused by a mutation in the gene responsible for the synthesis of beta-globin, leads to a change in the shape of red blood cells, which in turn causes their abnormal aggregation, impeding blood circulation and reducing the level of oxygen in the tissues. The relevance of the problem lies not only in the high prevalence of the disease, but also in the complexity of its diagnosis and treatment.

Keywords: Sickle cell anemia, clinical symptoms, laboratory diagnostics, treatment.

Introduction

Sickle cell anemia is an inherited blood disorder that occurs due to a mutation in the gene responsible for the synthesis of β-globin. The main reason is the replacement of adenine with thymine in codon 6 of the HBB gene, which leads to the formation of aberrant hemoglobin, known as hemoglobin S. When oxygen levels are low, such molecules clump together into aggregates, taking on a sickle-shaped shape, which makes red blood cells less elastic. which leads to congestion and oxygen starvation of tissues.

Other causes of the disease may include heredity, as sickle cell anemia is inherited in an autosomal recessive manner. Carriers of one copy of the mutated gene, known as "sickle cell cell carriers," may not show symptoms, but under conditions of stress, infection, or high altitudes above sea, health risks increase substantially [1, 8, 17].

Clinical symptoms of this condition include a number of manifestations, primarily anemia, which is manifested by fatigue, weakness and shortness of breath. Frequent crises of pain caused by blockage of small vessels by sickle cells become a characteristic feature. These crises can affect various organs, including the chest, abdomen and joints, causing intense pain [1, 5, 19].

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In addition, patients often have infections, especially pulmonary infections, due to insufficient function of the spleen. Skin manifestations, such as leg ulcers, can also be a consequence of chronic hypoxia and impaired circulation. Chronic anemia can lead to damage to organs such as the liver and kidneys, which requires close medical monitoring and a comprehensive approach to treatment, including regular transfusions and the use of drugs that stimulate the production of normal red blood cells.

Laboratory diagnosis of sickle cell anemia is based on the detection of specific changes in the blood associated with the presence of abnormal red blood cells. The main method is blood microscopy, which allows visualizing sickle cells, which have a characteristic shape that leads to impaired microcirculation and oxygenation of tissues [3, 12, 13].

Complete blood count (CBC) for sickle cell anemia is important for diagnosing and monitoring the patient's condition. In this disease, characteristic changes in blood counts are observed. The main sign is a decrease in hemoglobin levels, which leads to anemia. As a result, a complete blood count may show low values of red blood cells and hematocrit, as well as a high level of reticulocytes, which indicates a compensatory reaction of the bone marrow [1, 11, 14].

Despite anemia, the formed elements may remain within the normal range, but it is important to identify features, such as the presence of sickle red blood cells, which can be established using specialized research methods. In addition, the CBC may show signs of hyperleukocytosis and changes in platelets, indicating possible complications, including infections and blood clots.

In addition, molecular genetic methods are used to confirm the diagnosis and identify mutations in the HBB gene, which is responsible for the production of beta-globin.

Molecular genetic techniques play a key role in the study of sickle cell anemia, an inherited disease caused by a mutation in the HBB gene encoding hemoglobin beta chains. Modern technologies such as polymerase chain reaction (PCR) and next-generation sequencing make it possible to identify the mutations responsible for the disease with high accuracy [2, 10, 15].

These methods not only contribute to early diagnosis, but also open up new horizons for the development of targeted therapies. For example, the use of gene therapy based on CRISPR-Cas9 shows promising results in correcting mutations, which can significantly improve patients' quality of life.

In addition, molecular genetic studies can establish a genetic predisposition to the disease, which plays an important role in the strategy of individualized treatment and preventive measures.

The hemoglobin electrophoresis test for sickle cell anemia is an important diagnostic tool to determine the types of hemoglobin present in the patient's blood. During electrophoresis, the blood is separated into fractions depending on the charge of hemoglobin molecules, which makes it possible to detect the presence of hemoglobin S, which is responsible for the pathological condition.

This test not only confirms the diagnosis, but also helps to assess the severity of anemia, as well as the risk of complications. The clinical significance of electrophoresis is that it can also detect other forms of hemoglobin, such as hemoglobin C and D, which is important for determining a more accurate prognosis and choosing a treatment strategy [3, 9, 16].

Sickle cell anemia leads to a variety of complications, including chronic anemia, abdominal pain, organ damage, and an increased risk of infections. The prognosis for patients with SCD depends

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on many factors, including the extent of the disease, the availability of medical care, and compliance with doctors' recommendations.

Modern approaches to the treatment and management of the disease have significantly improved the prognosis. Methods such as chemotherapy, stem cell transplantation, and new biologics have helped many patients to improve their condition. However, it is important to note that early diagnosis and regular medical monitoring remain key aspects in the management of SCD [2, 6, 18].

Some patients achieve a normal life expectancy and a high quality of life thanks to competent treatment and family support.

The treatment of sickle cell anemia is a multifaceted process aimed at improving the quality of life of patients and reducing the incidence of complications. The main goal of therapy is to prevent crises associated with microthrombosis and maintain normal hematocrit levels. The mainstay of treatment is hydroxyuric acid and transfusion therapy, which help reduce the number of sickle cells and increase the level of normal red blood cells.

Findings

Thus, current research is aimed at understanding the mechanisms of the disease, as well as developing new therapeutic approaches, including genetic therapy and innovative methods for managing symptoms. Raising public and medical awareness about sickle cell anemia is becoming an important step towards improving diagnosis and access to necessary medical services.

Psychosocial support for patients and their families is equally important, contributing to adaptation to a difficult life with a chronic disease. Specialized rehabilitation and education programmes can significantly improve the quality of life and give hope for better health. Each individual direction in the treatment of sickle cell anemia creates a synergy that contributes to advances in this field of medicine.

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